



Short report

Genetic data of a Brazilian population sample (Santa Catarina) using an X-STR decaplex

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ABSTRACT

The study of X-chromosomal short tandem repeats (X-STRs) can complement the analysis of autosomal and Y-STRs. A decaplex system for the X-chromosome genetic markers, DXS8378, DXS9898, DXS7133, GATA31E08, GATA172D05, DXS7423, DXS6809, DXS7132, DXS9902 and DXS6789, was used to study a population sample of Santa Catarina, Brazil. 184 individuals (72 female and 112 male samples) were typed. DNA was amplified in a multiplex reaction and the automatic detection performed using capillary electrophoresis. Allele frequencies and some forensic parameters were calculated.

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1. Population

A total of 184 healthy unrelated individuals (72 females and 112 males), was autochthonous from Santa Catarina, Brazil.

2. Material and methods

2.1. DNA extraction and amplification

DNA was extracted from blood samples using the Chelex[®] method¹ and purified, using a modified organic phenol-chloroform-isoamylalcohol method. It was amplified according to the protocol established for GEP-ISFG collaborative study on X-STRs^{2,3} that allows co-amplification of 10 loci: DXS8378, DXS9898, DXS7133, GATA31E08, GATA172D05, DXS7423, DXS6809, DXS7132, DXS9902 and DXS6789. PCR was performed in a total volume of 10 µl containing 5 µl of 2× Qiagen Multiplex PCR Master Mix (Qiagen), 1 µl of 10× Primer Mix, 3.5 µl of distilled water and 0.5 ml of template DNA. For all primers, the final concentration in

the reaction was 0.2 µM. Thermocycling conditions, using a GeneAmp[®] PCR System 9700 (Applied Biosystems) were: an initial denaturation for 15 min at 95 °C, followed by 10 cycles of 30 s at 94 °C, 90 s at 60 °C, 60 s at 72 °C; and 20 cycles of 30 s at 94 °C, 90 s at 58 °C, 60 s at 72 °C; and a final extension of 60 min at 60 °C.

The amplified products were detected and separated by capillary electrophoresis using an ABI PRISM[®] 3100 Genetic Analyzer (Applied Biosystems). The internal size standard LIZ 500 (Applied Biosystems) was used. Fragment sizes were determined automatically using the Genescan[®] Analysis Software version 3.7 and genotyping performed through comparison with DNA control samples 9948 (Promega) and 9947A (Applied Biosystems) according to the recommendations of Szibor et al.⁴ The data for the loci DXS8378, DXS7423 and DXS7132 have been reported previously.⁵

2.2. Data analysis

Allele frequencies were calculated by the direct counting method. The Hardy–Weinberg equilibrium was tested using the exact test with the GENEPOP version 3.4 software package⁶ (Table 1). Several parameters of forensic interest were estimated by using the formulas proposed by Desmarais⁷ (Table 2). An unrooted UPGMA (unweighted pair group method using arithmetic mean) tree was

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Table 1Allele frequencies of 10 X-chromosome STRs in a population sample of Santa Catarina, Brazil ($n = 256$ chromosomes).

	DXS7423	DXS7132	DXS6789	GATA172D05	DXS9898	DXS6809	DXS8378	DXS7133	GATA31E08	DXS9902
6				0.125						
7								0.004	0.167	0.004
8				0.179				0.04	0.023	
8.3					0.191					
9				0.058	0.019		0.012	0.451	0.152	0.043
10				0.315	0.008		0.311	0.132	0.280	0.307
11		0.016		0.198	0.171		0.335	0.350	0.265	0.362
12		0.078		0.113	0.346		0.315	0.043	0.101	0.237
13	0.086	0.292		0.004	0.222		0.023	0.012	0.012	0.043
14	0.323	0.307	0.004	0.008	0.043		0.004	0.004		
15	0.393	0.226	0.070							
16	0.163	0.054	0.039							0.004
17	0.035	0.023	0.004							
18		0.004	0.008							
19			0.008							
20			0.346							
21			0.253			0.004				
22			0.202							
23			0.047							
24			0.004							
25			0.004							
26										
27						0.016				
28			0.008							
29						0.016				
30						0.043				
31						0.148				
32						0.117				
33						0.362				
34						0.179				
35						0.093				
36						0.016				
37						0.008				

drawn with TREEVIEW software⁸ using the genetic distances computed with the GDA package.⁹

3. Results and other remarks

Allele frequencies are shown in Table 1. The distribution was similar to that previously found in other populations in the Iberian Peninsula,¹⁰ and particularly in the Portuguese population¹¹ (Fig. 1). This finding is consistent with the known historical migrations from Portugal to Brazilian region of Santa Catarina. No deviations from the Hardy–Weinberg equilibrium were observed ($P \geq 0.005$; significant level after Bonferroni correction), with the exception of the DXS6789 locus (Table 2). The deviation from HWE for DXS6789 may result from the relatively small number of studied individuals. The forensic efficiency parameters are shown in Table 2. There were no marked differences

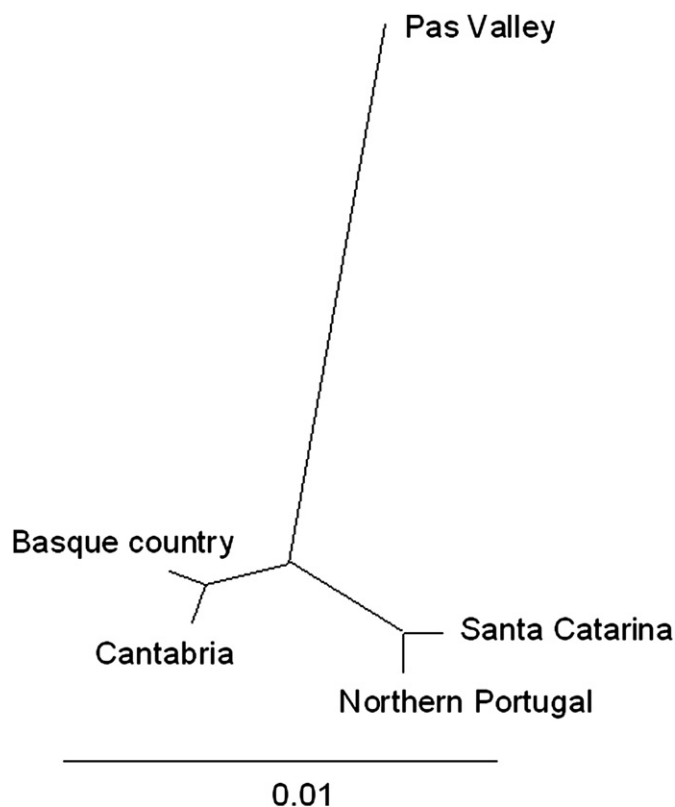
Table 2

Statistical parameters of forensic interest of the 10 X-chromosomal STRs in a population sample of Santa Catarina, Brazil (72 women and 112 men).

	<i>p</i>	PD-female	PD-male	PE-trio	PE-mo
DXS7423	0.9550	0.8626	0.7060	0.6551	0.5114
DXS7132	0.6669	0.9032	0.7596	0.7206	0.5852
DXS6789	0.0005*	0.9096	0.7646	0.7296	0.5964
GATA172D05	0.2413	0.9066	0.7629	0.7257	0.5904
DXS9898	0.6704	0.9066	0.7629	0.7257	0.5904
DXS6809	0.9590	0.9309	0.7902	0.7651	0.6392
DXS8378	0.9988	0.8409	0.6911	0.6275	0.4811
DXS7133	0.0151	0.8176	0.6541	0.5913	0.4455
GATA31E08	0.1465	0.9239	0.7896	0.7578	0.6289
DXS9902	0.6281	0.8663	0.7145	0.6623	0.5190

p: Hardy–Weinberg equilibrium exact test in the female sample; PD-female: power of discrimination in female cases; PD-male: power of discrimination in male cases; PE-trio: expected probability of exclusion in trio cases; PE-mo: expected probability of exclusion in motherless cases.

* Significant value after applying Bonferroni correction for multiple tests ($P \leq 0.005$).

**Fig. 1.** Unrooted UPGMA tree visualized with TREEVIEW software.

among loci, with power of discrimination estimates varying between 82% and 93%, for single locus analysis in female cases. Therefore, this is a robust decaplex that may be quite useful in kinship and identification cases, particularly when some ancestors are missing.

Conflict of interest

None declared.

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Ethical approval

None declared.

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